

Huntington disease Reference Materials Characterized by the GeT-RM

CAG Repeat Length as Determined by:

10 Clinical Genetic Laboratories

Sequencing
(1 Lab)
Mean³

Mean¹

Mode²

Allele 1 (95% confidence interval)	Allele 2 (95% confidence interval)	Allele 1 (95% confidence interval)	Allele 2 (95% confidence interval)	Allele 1/2	Cell Bank	DNA Number	Characterization methods ⁴ (# labs)
15 (13.7-15.8)	15 (13.7-15.8)	15 (77%)	15 (80%)	15/15	Coriell	NA20245	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
17 (15.9-18.3)	18 (17.2-19.1)	17 (77%)	18 (87%)	17/18	Coriell	NA20206	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
19 (17.7-20.7)	21 (20-22.1)	19 (67%)	21 (77%)	19/21	Coriell	NA20207	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
15 (14.1-15.9)	24 (22.3-25.9)	15 (80%)	24 (77%)	15/24	Coriell	NA20246	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
15 (14.1-15.9)	29 (28.1-29.9)	15 (80%)	29 (80%)	15/29	Coriell	NA20247	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
17 (15.0-19.3)	36 (35.1-37.3)	17 (77%)	36 (80%)	17/36	Coriell	NA20248	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
22 (21.1-22.8)	39 (38.3-39.9)	22 (87%)	39 (83%)	22/39	Coriell	NA20249	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
15 (14.1-15.8)	40 (39.2-41.0)	15 (83%)	40 (80%)	15/40	Coriell	NA20250	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
35 (33.4-36.3)	45 (43.5-46.5)	35 (80%)	45 (77%)	35/45	Coriell	NA20208	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
45 (43.8-46.2)	47 (46.3-47.8)	45 (73%)	47 (83%)	45/46	Coriell	NA20209	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
39 (38.1-40.0)	50 (49.1-50.8)	39 (80%)	50 (83%)	39/50	Coriell	NA20251	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
22 (21.2-22.9)	66 (63.7-67.5)	22 (83%)	65 (37%) 66 (37%)	22/65	Coriell	NA20252	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
17 (15.4-18.2)	74 (72.0-76.6)	17 (77%)	74 (50%)	17/75	Coriell	NA20210	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)
22 (20.3-23.3)	99 (95.8-102.7)	22 (73%)	100 (52%)	22/101	Coriell	NA20253	PCR-PAGE (5), PCR-CE (4), PCR-seq (1)

last updated 02-25-2008

¹Mean repeat length calculated from 30 responses per allele (Except for allele 2 of DNA sample NA20253 which had only 25 responses). Values were rounded to the nearest whole number. ²CAG repeat length reported most often out of 30 responses per allele (Except for allele 2 of DNA sample NA20253 which had only 25 responses), ³Unidirectional sequencing, results from one lab (National Institute of Standards and Technology)

⁴Methods: All labs used home-brew methods. Each lab amplified the CAG repeat region using PCR and separated the products using either polyacrylamide gel electrophoresis (PAGE), an ALF automated sequencer (seq) or capillary electrophoresis (CE). PCR product sizing was done manually (PAGE) or was automated (CE + seq).